EPP0813

Concomitant CNVs in healthy carriers with 7q31.1 microdeletions may suppress intellectual disability and autism spectrum disorders phenotype

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Introduction: About 66% of chromosomal microdeletions and microduplications associated with pathological conditions are inherited [Smajlagić D. et al., 2021]. The mechanisms of incomplete penetrance and variable expressivity of CNV are not fully understood. The presence of concomitant CNVs in the genome of healthy parents may have a modifying effect.
Objectives: Identification of additional CNVs in healthy carriers with 7q31.1 microdeletions.
Methods: CNVs were revealed by Agilent Technologies 60K microarray and confirmed by qPCR.
Results: We examined 3 families with inherited 7q31.1 microdeletions affecting only the *IMMP2L* gene, which is associated with intellectual disability, developmental delay and autism spectrum disorders. Family 1: Proband has intellectual disability, developmental delay, sensorimotor alalia. Microdeletion was inherited from the father, and a healthy sibling is also a carrier of rearrangement. In sibs, additional CNVs were identified: arr[hg19]: 4q31.21 (144722583_144939143)×3; 9p12p11.2(43588066_43836428)×3; 16p11.2(32066967_33773163)×1; and 17q21.31(44199517_44577208)×3. Family 2: Proband suffers from development delay, speech disorder and autism. Microdeletion was of paternal origin. The father additionally demonstrated microduplication 16p11.2p11.1(33967926-35204414)×3. Family 3: Proband was diagnosed with development delay and cerebral palsy. The mother is a carrier of a similar 7q31.1 microdeletion; two concomitant CNVs were identified in her karyotype: 9p13.1(39176840_40614884)×3; and 16p11.2p11.1(32833891_35204414)×3. Thus, healthy parents in 3 families have CNV in a common region 16p11.2, which contains the *TP53TG3* gene. It is important that *TP53TG3* expression is associated with epistatic CNV-CNV interactions [Sun, Kardia 2010].
Conclusions: Multiple CNVs in apparently healthy carriers of *IMMP2L* microdeltions may suppress disease phenotype due to the epistatic CNV-CNV interaction. This study was supported by Russian Science Foundation, grant no. 21-75-00112.

Disclosure: No significant relationships.
Keywords: intellectual disability; IMMP2L; CNV

EPP0812

Identification of candidate genes of intellectual disability by single-gene deletions/amplifications mapping using chromosomal microarray analysis

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Introduction: Disease-causing deletions/amplifications may include a single gene, several exons or single/part of exon, contributing to detection of novel pathogenic genes. The localization of single-gene deletion/amplification within the gene can affect its clinical manifestation.
Objectives: Improvement of diagnosis of intellectual disability.
Methods: aCGH with 60K Agilent microarrays, qPCR.
Results: Among 1099 patients with intellectual disability potentially pathogenic single-gene deletions/amplifications were detected in 51 individuals (5%). qPCR was used to verify aberrations in 21 patients (41%). Ten mutations were of maternal origin, four - paternal, two - *de novo*, another two were confirmed without analysis of parents, and three could not be confirmed. Single-gene aberrations involving the *AGBL4* (exon 2), *ASMT* (exon 9), *CYP2C18* (whole gene), *DDX10* (promoter, exons 1-13), *GYPA* (whole gene), *LIG4* (exon 1), *LSAMP* (intron 1), *PSD3* (promoter, exons 1-11), *SNTB1* (intron 1), *SPOCK3* (exons 6-12), *STAG2* (exons 7-34), *SYT10* (promoter, exons 1-2), *TCAF2* (exon 8), *TMPRSS15* (promoter, exons 1-12), and *ZDHHC7* (promoter, exons 1-4) genes were described by us for the first time. Deletion or amplification of several exons within a gene can affect transcription as point mutation does, while the copy number change of a whole gene can lead to an abnormal amount of the protein.
Conclusions: Fifteen novel genes potentially responsible for mental health were identified. In most of them aberrations were partial deletions/duplications. Most of abnormalities were inherited from healthy parents indicating the possible presence of a point mutation on the second allele or some modifying factors. This study was supported by the Russian Science Foundation, grant 21-65-00017.

Disclosure: No significant relationships.
Keywords: intellectual disability; Single-gene deletion; Single-gene amplification

EPP0813

Benefits of treadmill training for patients with Down Syndrome

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Introduction: Down syndrome (DS) is a complex condition that causes various health problems and it is accepted that treadmill training is a therapy method for some of them.
Objectives: The objective was to evaluate the effectiveness of various results of treadmill training in children and adults with DS.

Methods: We included studies in which participants with DS from every age group received treadmill training, alone or combined with physiotherapy and could optionally be compared to a control group with patients with DS who did not use treadmill training. The search was conducted in medical databases: PubMed, PEDro, Science Direct, Scopus and Web of Science and involved trials published until July 2021. Following PRISMA criteria, the Risk of Bias assessment was conducted using a tool developed by the Cochrane Collaboration for RCT. The included studies presented multiple outcomes and various methodologies therefore we were not able to conduct any sort of data synthesis, we presented measures of treatment effect as mean differences and corresponding 95% confidence intervals.
Results: 5 studies with a total number of 687 participants were included. 10 trials reported on walking onset, 8 on gait parameters or cardiovascular functions, 4 on anti-inflammatory effect and 3 on executive or cognitive functions. We came across 25 different outcomes in different age groups which are presented in a narrative manner. In all outcomes we have observed a positive result favouring the treadmill training.
Conclusions: Introducing treadmill exercise into typical physiotherapy generates improvements of mental and physical health of people with DS of all ages.

Disclosure: No significant relationships.
Keywords: treadmill training; intellectual disability; Down syndrome

Eating Disorders 02

EPP0819

Association of Eating Disorders with Recurrent Pain in Adolescent Girls

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Introduction: Eating disorders (ED) are associated with other mental illnesses, but the association of ED with pain is less well understood.
Objectives: To study the association of ED with headache and abdominal pain in adolescent girls.
Methods: In 2015-2018, 917 girls aged 12-17 were examined using the Body Image and Eating Distress scale (Koskelainen et al., 2001) and questions about frequency of recurrent headache and abdominal pain over the past six months. Adolescents were divided into three groups: girls with eating disorders (ED, n = 20); subthreshold eating disorders (SED, n = 88); and a control group (CG, n = 809).
Results: Headaches of varying frequency are were reported by 80% of girls with ED, 70.4% of girls with SED and 52.2% of CG girls. Frequent headaches (every week) were reported by 60% of girls with ED, 40.9% of girls with SED, and 29.9% of CG girls ($\chi2 = 20.21$, $p = 0.003$). Recurrent abdominal pain was reported by 65% of girls with ED, 56.8% of girls with SED, and by 46.6% of CG girls. Weekly abdominal pain affected 30% of girls with ED, 20.4% of girls with SED and 12.9% of CG girls. Combined weekly pain were commoner in girls with eating distress (in 20% of girls with ED and in 12.5% with SED) than in CG (7.7%, $\chi2 = 3.92$, $p = 0.04$).
Conclusions: Eating disorders in adolescents are often associated with pain, which can lead to late recognition of the disease, worsening its course and prognosis.

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Keywords: Adolescents; recurrent pain; Eating Disorders; girls

EPP0820

Suicidal Behavior in Adolescent Girls with Eating Disorders

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Introduction: Eating disorders are associated with suicidal behavior in adolescents. But the relationship between subthreshold eating disorders and suicidality is not well understood.
Objectives: To examine suicidal behavior in adolescent girls with eating disorders (ED) and subthreshold eating disorders (SED).
Methods: The study of 917 girls aged 12–17 used the Body Image and Eating Distress scale (Koskelainen et al., 2001) and questions about intentional self-harm (burns or cuts), suicidal thoughts and suicidal attempts. Adolescents were divided into three groups: girls with ED (n = 20); girls with SED (n = 88); and control group (CG, n = 809).
Results: Self-harm was reported by 55% of girls with ED, 35.2% of girls with SED and 20.2% of CG girls ($\chi2 = 15.82$, $p < 0.001$). Suicidal ideation was reported by 65% of girls with ED, 51.1% of girls with SED and 27.2% of CG girls ($\chi2 = 21.86$, $p < 0.001$). Suicidal attempts were reported by 45% of girls with ED, 17.1% of girls with SED and 7.5% of the CG. There were no differences in the prevalence of self-harm and suicidal ideation between the ED and SED groups, rates of suicide attempts were 2.6 times higher in the ED group.
Conclusions: Girls with both ED and SED have a high risk of suicidal behavior: 2.7-4.5 times higher rates of self-harm, 1.9-2.4 times higher rates of suicidal ideation, and 2.3-6 times higher rates of suicidal attempts. Management of such adolescents should include assessment of the risk of suicide.

Disclosure: The study was supported by the Russian Science Foundation grant # 21-15-00033.
Keywords: Eating Disorders; girls; Suicide; Adolescents